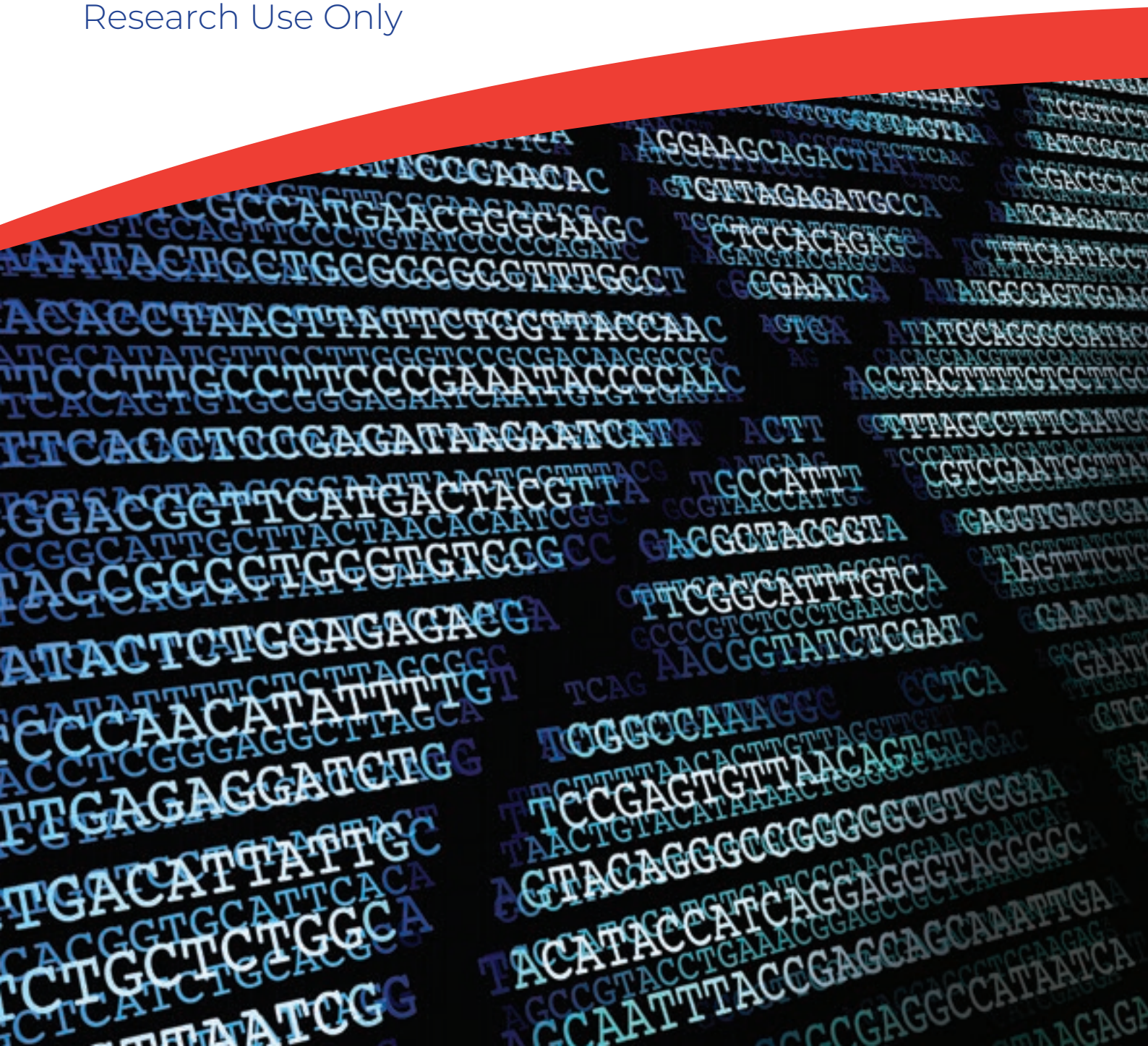
ALMAC

Partnering to Advance Human Health

Whole Genome Sequencing (WGS) Service

Research Use Only



Whole Genome Sequencing (WGS) Service

A high quality, robust Next Generation Sequencing (NGS) solution built on Roche's platform. Almac's wet lab optimised workflow generates comprehensive WGS data from blood. The platform covers coding and non-coding regions with readily interpretable raw sequencing data for use in biomarker discovery and retrospective clinical investigation.

Platform	
Intended Use	Research Use Only (RUO) for biomarker discovery and retrospective clinical investigation.
Technology	NGS Technology - Illumina sequencing platforms.
Type	Core laboratory service offering for WGS data generation with associated QC report.
Variants	Germline variants (SNPs, Insertions and Deletions).
Sample requirements	
Tissue type	Blood.
Recommended tissue requirements	Minimum 200ul EDTA Blood.
Input material requirements	Minimum recommended input of 250ng DNA from blood samples
Workflow	
Nucleic acid extraction	QIAamp DNA Blood Mini.
Library preparation	Roche KAPA HyperPrep (PCR-Free).
Platform performance	
Coverage	30x average on target coverage across cohort (sample quality dependent).
Quality control (QC)	
QC assessment	Comprehensive QC assessment of data including: · Sample & Library QC: DNA input, post-library preparation quality & quantity. · Sequencing Run QC. · Post Sequencing & Alignment QC: Including coverage QC and duplication rate.
Turnaround time	
TAT	For batch retrospective testing - turnaround time (TAT) agreed on a per project basis.
Reporting	
Raw data provided	FASTQ files.
QC Report	Excel compatible report, detailing lab, pre & post alignment QC. HTML interactive report allowing simultaneous assessment of sequence QC data from multiple samples.
Bioinformatics applications	Pre and post alignment QC. Germline variant discovery service including standard variant QC filtering and annotation steps. Pipelines hosted on cloud platform. Further downstream analysis & reporting can be facilitated by Almac Diagnostic Services Bioinformatics Team if required, at an additional cost.

Key benefits:

- **QC guarantee of 30X mean target coverage** across sample cohort ensures comprehensive downstream analysis and data interpretation.
- Protocol **suitable for blood samples.**
- **Interactive QC report** enables rapid and comprehensive sample quality interrogation across the cohort.
- **Raw Sequence data** delivered in FASTQ format, compatible with the majority of bioinformatics pipelines.
- **Germline variant discovery service:** filtered and annotated germline variants delivered as standard VCF files compatible with majority of downstream interpretation tools.

Performance validation:

To assess the robustness and performance of Almac's WGS Data Generation Platform, Almac performed a number of studies including input, accuracy, and repeatability assessments.

1. Input Study

The quality of WGS data generated at Almac using a range of high quality DNA derived from blood samples was assessed. Samples with 250ng and 500ng input generated data with an overall agreement of 99.99% (CI: 99.99-99.99).

250ng input vs 500ng input

Metric	Estimate	Confidence Interval
OPA	99.99	99.99 - 99.99
PPA	98.24	98.24 - 98.25
NPA	100.00	100.00 - 100.00

Conclusion – Minimum recommended input is 250ng DNA

2. Accuracy Study

WGS data generated at Almac using 250 ng of DNA isolated from Blood samples were compared to corresponding WES data generated at Almac for those same samples. Across all samples, the average on target coverage was 31X and average duplication rates 9.6%. Data demonstrated overall agreement at 99.99% (CI: 99.99-99.99). Additionally the commercial control (Genome in a Bottle control NA12878 (HG001)) demonstrated detection of >97% of the high-confidence variants.

WGS 250ng vs WES

Agreement Metric	Estimate	Confidence Interval
OPA	99.99	99.99 - 99.99
PPA	98.80	98.70 - 98.90
NPA	99.99	99.99 - 99.99

Conclusion - Highly accurate WGS data generation

3. Repeatability Study

The repeatability of WGS platform was assessed using DNA isolated from blood samples. Across all samples, repeatability estimates were greater than 99% for insertions, deletions and SNPs.

Variant Type	Estimate	Confidence Interval
Del	99.98	99.98 - 99.98
Ins	99.94	99.94 - 99.95
SNP	99.99	99.99 - 100.00
Combined	99.99	99.99 - 99.99

Conclusion - Highly reproducible and repeatable WGS data generation

Almac Diagnostics Services Expertise:

- More than 15 years' experience in working with challenging sample types, including FFPE.
- High quality service with robust quality control and data assurance.
- Data analysis and interpretation services available for customised investigation of data.
- Your study designed, managed & performed by PhD level scientists in our CLIA & CAP accredited laboratory.

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Not for use in diagnostic procedures.

Find out more:

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